

Case Report

Segmental neurofibromatosis

Virat Galhotra, Soheyl Sheikh¹, Sanjeev Jindal², Anshu Singla

Department of Pedodontics and Preventive Dentistry, ²Department of Oral Medicine and Radiology, Gian Sagar Dental College and Hospital, Vill, Ramnagar, Banur, Punjab, ¹Department of Oral Medicine and Radiology, M.M. College of Dental Sciences and Research, Mullana, Ambala, Haryana, India

Abstract

Segmental neurofibromatosis is a rare disorder, characterized by neurofibromas or café-au-lait macules limited to one region of the body. Its occurrence on the face is extremely rare and only few cases of segmental neurofibromatosis over the face have been described so far. We present a case of segmental neurofibromatosis involving the buccal mucosa, tongue, cheek, ear, and neck on the right side of the face.

Key words: Segmental neurofibromatosis, von Recklinghausen syndrome, neurofibromas

INTRODUCTION

Neurofibromatosis (NF) is an autosomal dominant disease, discovered in 1882, by the German pathologist Friedrich Daniel von Recklinghausen, characterized by disordered growth of ectodermal tissues, and is part of a group of disorders called Phakomatoses (neurocutaneous syndrome).^[1]

Neurofibromatosis type-I (NF-1), also known as von Recklinghausen syndrome, is caused by the mutation of a gene on the long arm of chromosome 17, which encodes a protein known as neurofibromin. The mutant gene is transmitted with an autosomal dominant pattern of inheritance. The incidence of NF-1 is about 1 in 3500 live births.^[1,2]

It is characterized by spots of increased skin pigmentation (café au lait spots), combined with peripheral nerve tumors and a variety of other dysplastic abnormalities of the skin, nervous system, bones, endocrine organs, and blood vessels.

The localized form of neurofibromatosis type-I, first described by Gammel in 1931, is very rare.^[3] Crowe *et al.* proposed the term sectorial neurofibromatosis for this localized form of neurofibromatosis, and

Miller and Sparkes modified the nomenclature to segmental neurofibromatosis (SN) — the current term for neurofibromas of segmental distribution.^[4] The commonly affected sites for SN are the thorax and abdomen (55%), upper extremities (20%), and lower limb and face (10% each). Only few cases (less than 10) of segmental neurofibromatosis over the face have been described so far.^[5]

CASE REPORT

A 26-year-old female patient reported to the Dental Department complaining about the unesthetic appearance of her face, since eight years [Figure 1]. History revealed that eight years back, the patient underwent surgery in the neck region for a cyst and one to two days after surgery the patient got a swelling on the face. The swelling was of the same size since then and there was no pain associated with the swelling. There was no history of difficulty in breathing or eating.

On examination, the face of the patient appeared asymmetrical, bilaterally, with a growth on the right side of the face. The skin over the right side of her face was thickened and hyperpigmented, with a soft and loose hanging overgrowth. There were numerous small and large, sessile and pedunculated nodular growths localized on the right side of her neck,

Access this article online

Quick Response Code:



Website:

www.ijdentistry.com

DOI:

10.4103/0975-962X.140847

Address for correspondence:

Dr. Virat Galhotra,
Department of Pedodontics and Preventive Dentistry,
Gian Sagar Dental College and Hospital, Vill. Ramnagar,
Banur - 140 401, Punjab, India.
E-mail: docvirat@gmail.com

with dark-colored underlying skin. The skin over the right cheek and chin area showed coffee-colored pigmentation (café-au-lait spots) [Figure 2].

There was a soft, compressible, nontender growth on the lower lip. The right ear showed disfigurement and hypertrophy, with thickened cartilage and overlying hyperpigmented skin [Figure 3].

Intraorally there were numerous soft tissue growths with a sessile base seen on the right buccal mucosa [Figure 4] and tongue [Figure 5]. The growths were soft, compressible, and nontender.

Laboratory findings revealed a raised erythrocyte sedimentation rate (ESR) (70 mm in the first hour), a decreased hemoglobin level (7.5 gm%), and the enzyme-linked immunosorbent assay (ELISA) was found to be nonreactive.

A panoramic radiograph disclosed asymmetric enlargement of the right side of the mandible, with

loss of cortication of the lower border in the midline area, expansion of the body of the mandible causing displacement of the teeth, seen on right side, expansion of the right inferior alveolar nerve canal, enlarged mental foramen, resorption of the roots of the mandibular right teeth, and loss of the right antegonial notch. A deficiency was seen on the right side of the maxilla, with crowding of the posterior teeth [Figure 6].

The right lateral oblique radiograph revealed a deep sigmoid notch, loss of the antegonial notch, an enlarged and rounded coronoid process, with resorption of the mandibular right teeth [Figure 7].

Subsequently, biopsy of one of the nodules was taken and sent for histopathological examination.

The histopathological slide revealed a non-encapsulated tumor of the dermis with a normal overlying epidermis. The tumor consisted of loosely spaced spindle cells and wavy collagenous strands in a clear matrix [Figure 8].



Figure 1: The unesthetic appearance of the face



Figure 2: Nodular growths localized on the right side of the neck with dark colored underlying skin



Figure 3: Disfigurement of the ear and hypertrophy with thickened cartilage and overlying hyperpigmented skin



Figure 4: Soft tissue growths with sessile bases on right buccal mucosa



Figure 5: Soft tissue growths with sessile bases on the tongue



Figure 6: A panoramic radiograph



Figure 7: A right lateral oblique radiograph

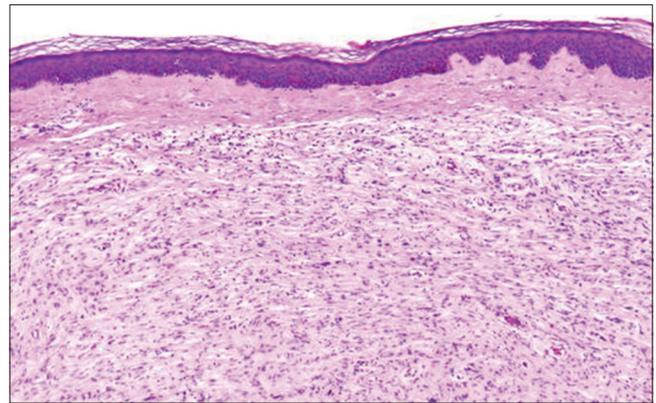


Figure 8: A non-encapsulated tumor of the dermis, with a normal overlying epidermis

DISCUSSION

Neurofibromas are the most common benign tumors of NF-1. These tumors are composed of Schwann cells, fibroblasts, mast cells, and vascular components. They can develop at any point along a nerve. There are three subtypes of neurofibromas: Cutaneous, subcutaneous, and plexiform.^[1,2]

Neurofibromatosis is often diagnosed because of unusual pigmentary patterns. The café au lait spots are irregularly-shaped, evenly pigmented, brown macules. Most individuals with neurofibromatosis have six or more spots that are 1.5 cm or greater in diameter. In young children, five or more café au lait macules, greater than 0.5 cm in diameter, are suggestive of neurofibromatosis. Less than 1% of the healthy children have three or more such spots, although one or two café au lait macules are commonly encountered in healthy individuals without disease.^[1-2,5]

Bone involvement includes pseudoarthrosis of the tibia, bowing of the long bones, and orbital defects. Localized bony hypertrophy, especially on the face, may be identified.

Treatment of neurofibromatosis is predominantly surgical. When neurofibromas increase in size or cause pain, malignant transformation must be suspected, and excision or biopsy must be performed. Acoustic neuromas and tumors that cause tinnitus and vertigo must be excised with great caution. Plastic surgeons may be included in the correction of deformities, especially those of the face. Considering the autosomal dominant inheritance pattern of neurofibromatosis, genetic counseling must be included in the treatment of all patients affected with this disease.^[1,5,6]

REFERENCES

1. Tinschert S, Naumann I, Stegmann E, Buske A, Kaufmann D, Thiel G, *et al.* Segmental neurofibromatosis is caused by somatic mutation of the neurofibromatosis type 1 (NF1) gene. *Eur J Hum Genet* 2000;8:455-9.
2. Roth RR, Martines R, James WD. Segmental neurofibromatosis. *Arch Dermatol* 1987;123:917-20.

3. Gammel JA. Localized neurofibromatosis. Arch Dermatol Syphilol 1931;24:712.
4. Miller RM, Sparkes RS. Segmental neurofibromatosis. Arch Dermatol 1977;113:837-8.
5. Koedijk FH. Segmental neurofibromatosis. Br J Dermatol 1987;117:790-1.
6. Loan JB, Fretzin DF, Bovenmyer DA. Genetic counseling in segmental neurofibromatosis. J Am Acad Dermatol 1990;22:461-7.

How to cite this article: Galhotra V, Sheikh S, Jindal S, Singla A. Segmental neurofibromatosis. Indian J Dent 2014;5:166-9.

Source of Support: Nil. **Conflict of Interest:** None declared.